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## Omphalocele, ten years experience at a tertiary care hospital

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### Abstract

Omphalocele is a rare congenital anterior abdominal wall defect. We present 12 cases of Omphalocele seen in our hospital. Further, we have discussed the clinical presentation, antenatal diagnosis, associated abnormalities and final outcome of the cases.

**Keywords:** Omphalocele, Abdominal wall, Antenatal diagnosis.

### Introduction

Omphalocele is a midline anterior abdominal wall defect with herniation of abdominal contents into the base of the umbilical cord. It may result from the failure of migration of lateral and cephalic or caudal folds during the fourth to sixth week after fertilization. Benson et al characterizes Omphalocele as the defects being greater than 4cm and extend to supraumbilical region, and distinguishing it from the herniation through the umbilical cord that has less defects than 4cm.<sup>1</sup> Bax et al used the presence or absence of liver in the sac to distinguish between a large and small Omphalocele.<sup>2</sup> It is commonly associated with other abnormalities particularly cardiac defects and aneuploidy.

Omphalocele is frequently seen in the context of congenital syndromes, with multiple associated anomalies, such as Beckwith Wiedemann syndrome, cloacal exstrophy, OEIS complex (Omphalocele, exstrophy, and imperforated anus, spinal), Cantrells pentalogy and trisomies. Cardiac abnormalities are described in 13%. Other chromosome abnormalities and pulmonary hypoplasia are also associated with these defects.

We present here a case series of 12 patients with Omphalocele to identify the clinical characteristics of foetal abdominal wall defects, managed at the setting and have provided information for the most likely clinical course of the affected fetuses.

### Case Report

This was a retrospective study carried out at the Aga Khan University Hospital, Karachi, a tertiary care hospital where the files of infants born with Omphalocele during a period of ten years from June 1999 to June 2009 were reviewed.

The pregnancies that were terminated due to Omphalocele were excluded from the study.

A performa was employed for the collection of data which included age at presentation, antenatal diagnosis, place of delivery, other associated findings, and the age and outcome of surgical intervention.

### Results

A total of 12 cases of Omphalocele were reported during the study period (Table). These included both Omphalocele major as well as minor. Out of these 12, one was delivered at home, while the rest were delivered at hospital. Out of 11, six were born outside AKUH and five were AKUH born. All were operated and discharged home. However, 1 case expired at age of 2 hours with multiple congenital abnormalities.

Out of the 12 cases, majority (n=11) cases clinically presented without ruptured Omphalocele, while only one was found to be ruptured.

Antenatal ultrasound was conducted in all the 12 cases, however only 3 were diagnosed to have Omphalocele. Furthermore, associated abnormalities were found in only 2 cases, one with bladder exstrophy, and the other with multiple congenital anomalies.

Six cases required ventilator support. The range of support given was from 2 hours to 1 month. Surgery was



Figure-1: One day Baby on ventilator after repair of giant Omphalocele.

**Table: Clinical Characteristic and Outcome.**

S. No	Clinical Presentation	Antenatal Diagnosis	Place of delivery	Any associated abnormality	Ventilation	Age at Surgery	Outcome
1	not ruptured	no	AKUH	no	24 hr	1 day	alive
2	not ruptured	no	outside	no	48 hrs	2 day	alive
3	not ruptured	yes	outside	no	24 hrs	3rd day	alive
4	not ruptured	no	home delivery	no	no	8 month	alive
5	not ruptured	no	outside	no	no	1 day	alive
6	not ruptured	yes	AKUH	no	1 month	1 day	alive
7	Ruptured	no	Outside	multiple	2 hr	not done	died
8	not ruptured	no	outside	no	24 hrs	5 day	alive
9	not ruptured	no	outside	no	no	7 day	alive
10	not ruptured	no	AKUH	no	no	1 day	alive
11	not ruptured	no	outside	bladder exstrophy	no	1 month	alive
12	not ruptured	yes	AKUH	no	no	1 day	alive

AKUH: Aga Khan University Hospital.



Figure-2: At one month of age, extubated and taking direct breast feeding.

conducted in 11 cases, with 6 having undergone the procedure on the first day. The most delayed surgery was conducted at 8 months after delivery. It was the same case that was delivered at home. Case-wise details are shown in the Table. Figure-1 and 2 show two pictures of two of the cases.

## Discussion

In literature, many studies have reported the incidence of Omphalocele to be 2.5 per 10,000 births.<sup>3</sup> However, varying incidence rates have been reported in different regions. A ten year review from Singapore reported the prevalence of Omphalocele to be 2.17 per 10,000 births.<sup>4</sup> In the study, the research has included very small number of cases, so the true prevalence is difficult to assess.

It has been known that ultrasound is an effective method for detection of Omphalocele.<sup>5</sup> Barisic et al evaluated 19 European registries and reported that sensitivity of ultrasound for detection of Omphalocele was 75%.<sup>6</sup> However, in the study, it was detected in only three cases out of 12 cases, giving the sensitivity to be 25%. In the setup, this series most of the deliveries were carried out in peripheral hospitals where expertise for ultrasounds was limited. Omphaloceles are also difficult to detect on antenatal ultrasound.

Clausener et al<sup>7</sup> recently carried out a 25 years review of 132 patients with anterior abdominal wall defects. They commented on improved results of surgery following the advent of prenatal diagnosis, partly due to selected abortion of more severe cases with multiple abnormalities. They have concluded that development and quality of life are not significantly reduced after survival of an isolated defect. A study from Peshawar, Pakistan reported conservative treatment by using mercurochrome (0.5-1%) solution of giant Omphalocele to be very effective. It was followed by delayed primary closure.<sup>8</sup> Thus, such measures are effective in areas where standard care units are not available.

About 80% of prenatally diagnosed Omphalocele are delivered by caesarean section, allowing the Omphalocele sac to be better protected from rupture.<sup>9</sup> We showed that Caesarean section was done in six cases (50%). Nevertheless, several studies have shown no difference in outcome between vaginal and caesarean delivery. In one study from Germany amniocentesis was carried out in 54% in the first half of pregnancy and the karyotype was normal in all but one case.<sup>10</sup> In our study it was done in only one case (8%).

## Conclusion

Omphalocele is although rare, however remarkable results are achieved if it is properly diagnosed and managed. Radiological findings play an important role and efforts must be made to critically ensure the detection of such condition in infants.

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